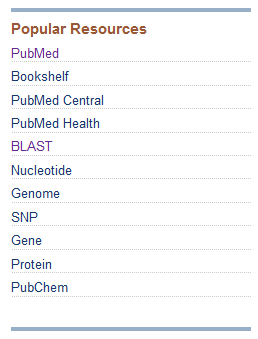
**Introduction To Bioinformatics:**

**Exercise 1:**

You will be given a nucleotide sequence found in real human DNA that is associated with a genetic disease when mutated. Your job is to compare the sequences you are given with the nucleotide sequence of most known genes, using the BLAST tool to search genetic databases.

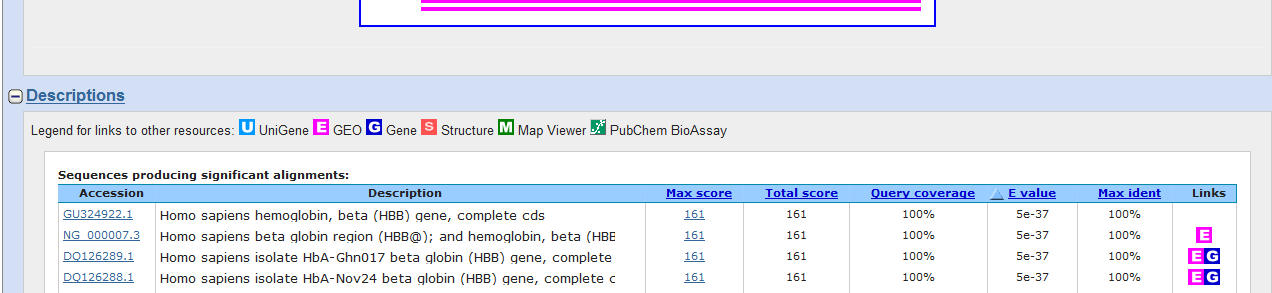
1. Go to the homepage for the NCBI ( [www.ncbi.nlm.nih.gov](http://www.ncbi.nlm.nih.gov) )
2. Click on the word "BLAST" located in the list of links on the right of the page.
3. Scroll down until you find the heading "**Nucleotide BLAST**" and click the link.



1. This time we will practice using a part of the gene sequence that codes for hemoglobin. When a mutation occurs in this gene a person can wind up with Sickle Cell Anemia. Cut and Paste the sequence below into the blast window.

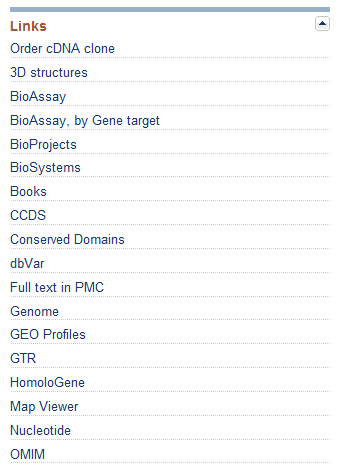
**GGG ATG AAT AAG GCA TAT GCA TCA GGG GCT GTT GCC AAT GTG CAT TAG CTG TTT GCA GCC TCA CCT TCT TTC ATG GAG TTT AAG ATA**



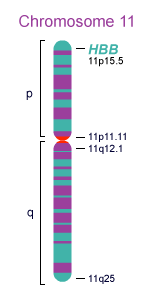
1. When you have finished entering your sequence, click the radio button for OTHERS and then click on the BLAST button.
2. You will then see a screen asking you to wait 10-20 seconds. Don’t click on anything just relax and wait patiently for the search to conclude.
3. After the search has ended, scroll down past the box with all of the pink bars in it and find the words **"Sequences Producing Significant Alignment".**
4. Listed in order are the closest matches with your DNA sequence. You should notice that the blast search will return results for all genomes currently mapped (Several prokaryotes, humans, rats, chimpanzees, cows, pigs, chickens and puffer fish to name a few)
5. Please take a moment to be awed by the similarity found in the DNA code despite the outward physical diversity of organisms.
6.  Click on the blue reference number (right side) attached to the second match. This will tell you the name of the gene and its abbreviation if available).
7. Scroll down to “Related Info” on the right side and click “Gene”

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1. This is the Gene Summary. It tells you that this sequence is officially recognized. HBB is the official Symbol for this gene. If you read further it will tell you that it is found in humans and that a mutated version of it causes Sickle Cell Anemia.
2. On the right you will see a link to PHENOTYPES. Click the link and look for information about Hb SS Disease under summary from GeneReviews. This will open more information about sickle cell.



1. Return to the Gene Summary page. Next click on the link to the “Map Viewer” in the right column under the “Related Sites” heading.



1. Map Viewer page. Across the top of the page, you will notice the numbers 1-22 XY. These numbers represent the chromosomes found in humans.
2. While we are here lets take a moment to go over chromosomal nomenclature

**11p15.5–** This means that the HBB gene is found on chromosome 11 on the short (p) arm in region 15.5. the fancy word for this is Locus which is science talk for location.



1. Your next step will be to click on the OMIM link in the information bar. This will take you to a page with lots of information about your gene and what it does.